

Note: The following information is provided by the author(s) and has not been reviewed by GeneReviews staff.

Table 2. *LGI1* Mutations Reported as of January 2007

Exon	Mutation (numbered from initiation site)	Effect on protein	Reference
1	124T>C	C42R	Ottman et al 2004
1	124T>G	C42G	Berkovic, Izzillo et al 2004
1	136T>C	C46R	Gu, Brodtkorb et al 2002, Pizzuti et al 2003
3	329C>A	A110D	Ottman et al 2004
3	329delC	Frameshift; protein truncation	Hedera et al 2004
3-4	IVS3(-3)C>A	Intron retention; protein truncation	Kalachikov et al 2002
4-5	IVS4(+1)G>A	Exons 3 and 4 skipped in the transcript; 48 aa truncated	Chabrol et al 2007
5	435C>G	S145R	Hedera et al 2004
5	461T>C	Leu154Pro	Pisano et al 2005
6	598T>C	C200R	Michelucci et al 2003
6	611delC	Frameshift; protein truncation	Kalachikov et al 2002
7	695T>C	L232P	Chabrol et al 2007
7	758delC	Frameshift; protein truncation	Morante-Redolat et al 2002
7-8	IVS7(-2)A>G	Intron retention; protein truncation	Kobayashi et al 2003
8	953T>G	F318C	Fertig et al 2003
8	893T>C	I298T	Ottman et al 2004
8	1050- 1051delCA	Frameshift; protein truncation	Kalachikov et al 2002
8	1148A>C	E383A	Kalachikov et al 2002
8	1295T>A	V432E	Michelucci et al 2003
8	1418C>T	Ser473Leu	Berkovic, Izzillo et al 2004
8	1420C>T	Premature stop codon; protein truncation	Morante-Redolat et al 2002; Bisulli, Tinuper, Scudellaro et al 2004 <i>(de novo)</i>
8	1639-1640insA	Frameshift; protein truncation	Kalachikov et al 2002

